

**WHAT IS CLAIMED IS:**

1           1. An isolated nucleic acid and the degenerate  
2. sequences thereof, which encodes human  $\alpha 1$  chain collagen  
3 protein, comprising the nucleotide sequence set forth in SEQ  
4 ID NO. 5.

1           2. The nucleic acid as claimed in claim 1, wherein the  
2 human  $\alpha 1$  chain collagen protein encoded by the nucleic acid  
3 has the amino acid sequence set forth in SEQ ID NO. 1.

1           3. The nucleic acid as claimed in claim 2, wherein the  
2 human  $\alpha 1$  chain collagen protein comprises:  
3           (i) von Willebrand factor A domain set forth in SEQ ID  
4 NO. 2;

5           (ii) thrombospondin N-terminal-like domain set forth in  
6 SEQ ID NO. 3; and

7           (iii) collagenous domain set forth in SEQ ID NO. 4.

8           4. The nucleic acid as claimed in claim 1, comprising  
9 DNA and RNA.

1           5. The nucleic acid as claimed in claim 4, wherein the  
2 DNA comprises cDNA and genomic DNA.

1           6. A human  $\alpha 1$  chain collagen protein having the amino  
2 acid sequence set forth in SEQ ID NO. 1.

1           7. The human  $\alpha 1$  chain collagen protein as claimed in  
2 claim 6, wherein the protein is encoded by the nucleic acid  
3 of claim 1.

1           8. The human  $\alpha 1$  chain collagen protein as claimed in  
2 claim 7, comprising:

3           (i) von Willebrand factor A domain set forth in SEQ ID  
4 NO. 2;

5           (ii) thrombospondin N-terminal-like domain set forth in  
6 SEQ ID NO. 3; and

7           (iii) collagenous domain set forth in SEQ ID NO. 4.

1           9. A recombinant vector comprising the nucleic acid of  
2 claim 1 and a regulatory sequence.

1           10. The recombinant vector as claimed in claim 9,  
2 wherein the regulatory sequence comprises an operatively  
3 linked promoter.

1           11. The recombinant vector as claimed in claim 9,  
2 wherein the recombinant vector is designated Bluescript  
3 KS(+)/*E. coli* DH5 $\alpha$ (*hCOLA1*) and deposited at the Culture  
4 Collection and Research Center (Hsinchu, Taiwan) and  
5 assigned accession number CCRC 940331.

1           12. A method for producing human  $\alpha 1$  chain collagen  
2 protein, comprising the steps of:

3           (a) transforming or transfecting a host cell with the  
4 recombinant vector of claim 9;

5           (b) culturing said transformed or transfected cell  
6 under the conditions sufficient for expression of the human  
7  $\alpha 1$  chain collagen protein; and

8           (c) recovering and purifying the human  $\alpha 1$  chain  
9 collagen protein.

1           13. The method as claimed in claim 12, wherein the host  
2 cell is selected from the group consisting of prokaryotic  
3 and eukaryotic cell.

1           14. The method as claimed in claim 13, wherein the  
2 prokaryotic cell comprises *Escherichia coli*.

1           15. The method as claimed in claim 13, wherein the  
2 eukaryotic cell comprises mammalian cell.

1           16. The method as claimed in claim 12, wherein the  
2 recovering and purifying step is conducted by column  
3 chromatography.

1           17. An isolated nucleic acid comprising at least 500  
2 contiguous nucleotides in length derived from SEQ ID NO. 5  
3 or a complementary nucleotide sequence thereto.

1           18. A kit for detecting the disease related to the  
2 mutation of SEQ ID NO. 5 in a mammal or human comprising a  
3 probe, which comprises the nucleic acid of claim 1 or claim  
4 17.

1           19. An isolated nucleic acid comprising at least 20  
2 contiguous nucleotides in length derived from SEQ ID NO. 5  
3 or a complementary nucleotide sequence thereto.

1           20. A kit for detecting the disease related to the  
2 mutation of SEQ ID NO. 5 in a mammal or human comprising a  
3 primer, which comprises the nucleic acid of claim 19.